malonyl-CoA decarboxylase deficiency

Malonyl-CoA decarboxylase deficiency is a condition that prevents the body from converting certain fats to energy. The signs and symptoms of this disorder typically appear in early childhood. Almost all affected children have delayed development. Additional signs and symptoms can include weak muscle tone (hypotonia), seizures, diarrhea, vomiting, and low blood sugar (hypoglycemia). A heart condition called cardiomyopathy, which weakens and enlarges the heart muscle, is another common feature of malonyl-CoA decarboxylase deficiency.

Frequency

This condition is very rare; fewer than 30 cases have been reported.

Genetic Changes

Mutations in the *MLYCD* gene cause malonyl-CoA decarboxylase deficiency. The *MLYCD* gene provides instructions for making an enzyme called malonyl-CoA decarboxylase. Within cells, this enzyme helps regulate the formation and breakdown of a group of fats called fatty acids. Many tissues, including the heart muscle, use fatty acids as a major source of energy.

Mutations in the *MLYCD* gene reduce or eliminate the function of malonyl-CoA decarboxylase. A shortage of this enzyme disrupts the normal balance of fatty acid formation and breakdown in the body. As a result, fatty acids cannot be converted to energy, which can lead to characteristic features of this disorder including low blood sugar and cardiomyopathy. Byproducts of fatty acid processing build up in tissues, which also contributes to the signs and symptoms of malonyl-CoA decarboxylase deficiency.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- deficiency of malonyl-CoA decarboxylase
- malonic aciduria

- malonyl-coenzyme A decarboxylase deficiency
- MCD deficiency

Diagnosis & Management

Formal Diagnostic Criteria

- ACT Sheet: Elevated C3-DC acylcarnitine
  https://www.ncbi.nlm.nih.gov/books/NBK55827/bin/C3-DC.pdf

Genetic Testing

- Genetic Testing Registry: Deficiency of malonyl-CoA decarboxylase
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0342793/

Other Diagnosis and Management Resources

- Baby's First Test
  http://www.babysfirsttest.org/newborn-screening/conditions/malonic-acidemia

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Health Topic: Genetic Brain Disorders
  https://medlineplus.gov/geneticbraindisorders.html
- Health Topic: Lipid Metabolism Disorders
  https://medlineplus.gov/lipidmetabolismdisorders.html
- Health Topic: Newborn Screening
  https://medlineplus.gov/newbornscreening.html

Genetic and Rare Diseases Information Center

- Malonyl-CoA decarboxylase deficiency
  https://rarediseases.info.nih.gov/diseases/3371/malonyl-coa-decarboxylase-deficiency

Educational Resources

- Disease InfoSearch: Malonyl-CoA decarboxylase deficiency
  http://www.diseaseinfosearch.org/Malonyl-CoA+decarboxylase+deficiency/4439
- MalaCards: malonyl-coa decarboxylase deficiency
  http://www.malacards.org/card/malonyl_coa_decarboxylase_deficiency
- Orphanet: Malonic aciduria
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=943

Patient Support and Advocacy Resources

- Children Living with Inherited Metabolic Diseases (CLIMB) (UK)
  http://www.climb.org.uk/
- Organic Acidemia Association
  http://www.oaanews.org/ma.html
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/metaboli.html

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22malonyl-coa+decarboxylase+deficiency%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28malonyl-coa+decarboxylase+deficiency%5BTIAB%5D%29+OR+%28malonic+aciduria%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- MALONYL-CoA DECARBOXYLASE DEFICIENCY
  http://omim.org/entry/248360

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